Are patients with truncal type of Stiff Person Syndrome hidden in a heterogeneous group of non-radiographic axial spondyloarthritids?

Sirs,

We have recently published a paper entitled “Extremely rare coincidence of non-radiographic axial spondyloarthopathy HLA-B27 positive and stiff person syndrome – rheumatologist point of view” in which we were the first to describe such a coincidence (1). In the three months following the publication of our case report, we received e-mails from two patients in the USA who had been treated as non-radiographic HLA B27 positive spondyloarthritids for 10 years until they had been diagnosed with anti-GAD positive stiff person syndrome (SPS). Their story is similar to our patient’s and we believe that HLA B27 positive in truncal form of stiff person syndrome may not be as rare as we thought. It seems that there is an entire subgroup of patients with non-radiographic axial spondyloarthritids (axSpA) that are B27 positive and have SPS.

Non-radiographic spondyloarthritids that do not involve radiographic changes of the sacroiliac joints was acknowledged as a variant of axial spondyloarthritids (axSpA) in 2009 by Assessment of Spondyloarthritids International Society (ASAS). Besides non-radiographic spondyloarthritids, these criteria include ankylosing spondylitis (that fulfils the modified New York criteria) in the axSpA group (2). A paper was published on national prevalence of axSpA in the USA in 2010. The prevalence estimates of ankylosing spondylitis and non-radiographic axial SpA are 0.35%, respectively (3). Although there are clinical similarities between patients with axSpA, a growing number of papers testify to the heterogeneity of non-radiographic axial spondyloarthritids. Furthermore, these papers emphasise the need for further observation of non-radiographic axSpA in order to define the disease, the treatment and the prognosis more precisely (4-6).

SPS is a rare autoimmune neurological disorder. It is characterised by progressive stiffness and rigidity and the prominence of truncal muscles accompanied with co-contraction of agonist and antagonist muscles. Its prevalence is less than 1 in a million individuals. Auto-antibodies against glutamic acid decarboxylase (GAD) are present in 80% of all patients (7), while amphiophysine antibody is present in fewer patients and is connected to cervical stiffness (8).

Since we are now aware of three cases of HLA B27 positive of non-radiographic axSpA and SPS, we think that in the group of patients with the diagnosis of non-radiographic axSpA HLA-B27 positive, an additional anti-GAD, amphiophysine antibodies together with EMG of paravertebral muscles should be made to exclude SPS. We presume that HLA typisation of truncal form of SPS might show high HLA-B27 incidence. However, there is a possibility that non-radiographic axSpA is misdiagnosed in some patients with truncal rigidity and that those patients have unrecognised SPS.

Related to that, attention should be focused on patients that do not react well to disease-modifying anti-rheumatic drugs (especially anti-TNF drugs) and in whom stiffness of paravertebral muscles persists.

Our dilemma is best summed up in the question: is it really a coincidence of two diseases or were the patients with Stiff-Person syndrome misdiagnosed from the start?

In order to answer our question, we have started a multicenter research trial in which all the patients with non-radiographic axSpA and SPS will be included and we will try to compare their clinical, radiological and laboratory findings. Our plan is to collect more data about these conditions that will help physicians in the early detection and differential diagnosis of these two autoimmune diseases. Moreover, we are willing to cooperate with other medical centres, which also are considering that the real nature of this new axSpA group is still not familiar enough. Since there is a significant heterogeneity of clinical and radiological presentation of axSpA, long-term studies are needed to better understand this disease and to achieve greater homogeneity (6).

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References
1. MARINOVIĆ I, PIVALICA D, ALJINOVIĆ J, VLAK T, ŠKORIC E, MARTINOVIĆ KALITERNA D: Extremely rare coincidence of non-radiographic axial spondyloarthropathy HLA-B27 positive and Stiff Person Syndrome – rheumatologist point of view. Mod Rheumatol 2013 Dec 2 [Epub ahead of print].